Aneuploidy and Structural Aberrations in Sperm of Healthy Men Detected by Multicolor FISH with Probes for Chromosomes 1, 16, 21, X, and Y.

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Numerical and structural chromosomal aberrations have important effects on the viability and health of the human embryo and neonate. The paternal contribution to aneuploidy ranges from about 10 to 100% depending on the specific chromosome involved. Two multicolor FISH assays (X-Y-21 and A-M-16) were applied to detect aneuploidy involving chromosomes 1, 16, 21, X, and Y and structural aberrations involving chromosome 1. Sex ratios were not significantly different from 1:1 among 63,582 sperm from four healthy donors. The frequencies of sperm disomic for chromosome 1, 16, and 21 were 5.0, 3.5, and 6.9 per 10^4 nuclei, respectively. These disomy frequencies were not statistically different from those established previously by the X-Y-8 and the A-M-8 FISH assays for the same donors. Furthermore, the frequency of sperm disomic for chromosome 21 was similar to that determined in previous studies using the hamster-egg technique: 44 per 63,582 (FISH) vs. 5 per 5,997 (hamster technique) ($p = 0.7$). The hyperhaploidy frequency of the sex chromosomes was 15.4 per 10^4 nuclei, while the diploidy frequencies in both assays were approximately 16 per 10^4 nuclei. Loss and gain of chromosome arm 1p was detected in 5.5 per 10^4 nuclei, which was comparable with data for the same men using the previous A-M-8 assay. The calculated percentage of sperm carrying chromosomal imbalance was 1.4%, which was similar to the percentage of a subgroup of structural aberrations evaluated by the hamster-egg technique. These findings provide initial validation for the aneuploidy frequencies and structural aberration rates obtained with the X-Y-21 and A-M-16 sperm FISH assays. Future studies will investigate the effects of exposure to toxicants and the persistence of damage after the end of exposure. [Work was performed under the auspices of the US DOE by the Lawrence Livermore Natl. Lab. under contract W-7405-ENG-48; A.B. was supported by EU Contract EV5V-CT94-0403 and the US DOE]